ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet

[Elevated C0/C16+C18 Acylcarnitine]

Carnitine Palmitoyltransferase I Deficiency (CPT I)

Differential Diagnosis: Carnitine palmitoyltransferase I (CPT I); carnitine supplementation.

Condition Description: Carnitine palmitoyltransferase I (CPT I) deficiency disrupts an early component of the carnitine shuttle, the system used to bring long-chain fatty acids into mitochondria for energy production. A deficiency of CPT I activity results in the accumulation of free carnitine and the inhibition of the fatty acid oxidation response necessary to generate energy during fasting and periods of increased energy needs, such as fever or stress. Presentation in the neonatal period is rare.

You Should Take the Following IMMEDIATE Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (lethargy, seizures).
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn (lethargy, hepatosplenomegaly, or seizures). If any of these findings are present or if the newborn is ill, transport to a hospital for further treatment in consultation with the metabolic specialist.
- Initiate confirmatory/diagnostic testing and management, as recommended by specialist.
- Provide the family with basic information about CPT I and its management.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: **Plasma carnitine (free and total):** Free carnitine is elevated in CPT I deficiency. **Dried blood spot or plasma acylcarnitines:** Free carnitine is elevated and long-chain acylcarnitines are decreased or are normal. **Molecular genetic testing** may be required to establish the diagnosis.

Clinical Considerations: CPT I deficiency can have a variable presentation. Critical hypoketotic hypoglycemia leading to lethargy, hepatomegaly, and seizures is a common presenting feature usually precipitated by fasting or acute illness; this presentation is rare in newborns. The Arctic *CPT1* variant is common among the Inuit population of Alaska, Canada, and Greenland. Most infants with this variant remain asymptomatic; those who do develop symptoms usually do so only early in life during periods of fasting or acute illness.

Additional Information:

How to Communicate Newborn Screening Results
Emergency Protocols (New England Consortium of Metabolic Programs)
Gene Reviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse

Referral (local, state, regional, and national):

Find a Genetics Clinic Directory
Genetic Testing Registry

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State and Other Resources

State Newborn Screening Program

Newborn Genetic Screening, New Mexico Department of Health
877-890-4692, www.nmhealth.org/about/phd/fhb/cms/nbgs/

Genetics/Metabolic Consultants

Michael Marble, MD
University of New Mexico Metabolic Clinic
505-272-6631, hsc.unm.edu/directory/marble-michael.html

Information for Clinicians and Families

New Mexico Home Portal (see Newborn Disorders and Parents & Families sections)
nm.medicalhomeportal.org/newborn/carnitine-palmitoyltransferase-1a-deficiency

Parent/Family Support

FOD Family Support Group
fodsupport.org/

National Resources (with web addresses)

Additional Information

How to Communicate Newborn Screening Results
www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Emergency Protocols (New England Consortium of Metabolic Programs)
https://www.newenglandconsortium.org/cpt-i-deficiency

Gene Reviews
https://www.ncbi.nlm.nih.gov/books/NBK1527/

Medline Plus
https://medlineplus.gov/genetics/condition/carnitine-palmitoyltransferase-i-deficiency/

Condition Information for Families-HRSA Newborn Screening Clearinghouse
https://newbornscreening.hrsa.gov/conditions/carnitine-palmitoyltransferase-i-deficiency

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
clinics.acmg.net

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/